Variant: NM_005633.3(SOS1):c.2988G>A (p.Pro996=)

CA136123

40713 (ClinVar)

Gene: SOS1 (HGNC:6654)
Condition: RASopathy (MONDO:0021060)
Inheritance Mode: Autosomal dominant inheritance
UUID: 47edbaff-50ac-42ee-838b-2ac0c4a4054c
Approved on: 2017-04-18
Published on: 2018-12-10

HGVS expressions

NM_005633.3:c.2988G>A
NM_005633.3(SOS1):c.2988G>A (p.Pro996=)
NC_000002.12:g.38997015C>T
CM000664.2:g.38997015C>T
NC_000002.11:g.39224156C>T
CM000664.1:g.39224156C>T
NC_000002.10:g.39077660C>T
NG_007530.1:g.128449G>A
ENST00000395038.6:c.2988G>A
ENST00000402219.6:c.2988G>A
ENST00000426016.5:c.2988G>A

Benign

Met criteria codes 1

BA1

Evidence Links 0

Expert Panel

RASopathy VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

RASopathy VCEP

The filtering allele frequency of the c.2988G>A (p.Pro996=) variant in the SOS1 gene is 0.396% (51/10072) of African chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Met criteria codes

BA1	✓	The filtering allele frequency of the c.2988G>A (p.Pro996=) variant in the SOS1 gene is 0.396% (51/10072) of African chromosomes by the Exome Aggregation Consortium, which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen RASopathy Expert Panel (BA1; PMID:29493581)

Curation History

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